What is Fragile X Syndrome?

Fragile X syndrome is a genetic condition that occurs in people of all racial and ethnic backgrounds. It is more common in males, but also affects females. Fragile X syndrome is a developmental disorder that affects learning and behavior. It occurs in about 1 in 2,000 people.

How may Fragile X syndrome affect my child?

Learning: Early motor skills, speech, and language development are commonly delayed in childhood. People with fragile X syndrome usually have a learning disability. The degree may range from mild impairment to severe mental retardation. Females are often less severely affected than males.

Behavior: Behavioral problems are common in both males and females. These may include unusual hand movements, hyperactivity, anxiety, autism spectrum disorder, and other behaviors.

Physical: Certain facial features may be noticed by a medical professional. These include a long face, prominent forehead and chin, and large ears. Head size may be larger than average. The joints may be especially flexible. After puberty, large testes are common in males.

Medical: Lazy eye, curvature of the spine (scoliosis), seizures (epilepsy) or heart murmurs occur in some people with Fragile X. Women who carry a certain genetic change may have early menopause. Older adults who are carriers may develop a condition that causes tremors and affects balance and memory, similar to Parkinson's disease.

How does Fragile X syndrome occur?

Fragile X syndrome is caused by a change (mutation) in a gene on the "X" chromosome. Fragile X syndrome may run in a family. Women who carry the mutation, or a smaller change called a "premutation", are more likely to have affected children. Men who carry the premutation are not expected to have affected children, but their grandchildren (daughter's children) may have Fragile X syndrome. The genetics of Fragile X syndrome is complex. Genetic counseling is recommended for parents to learn more about Fragile X syndrome in their family, and possible risks for their children.

How is Fragile X syndrome treated?

Fragile X syndrome cannot be cured, but some symptoms can be treated. Infants and toddlers (birth to 3 years) should be connected with *Early On*® Michigan as soon as possible. If there are concerns about learning, speech, or behavior in a child over 3 years of age, a referral for special education services should be made. Children often benefit from physical and occupational therapy, sensory integration, and tools to assist with communication. Other therapies or treatments may be needed for behavioral or health problems as they arise. Children with Fragile X syndrome and their families benefit from having a primary care physician who helps to coordinate their care with medical specialists and other community-based services.

For more information, call Michigan's Genetics & Birth Defects Program toll-free at 1-866-852-1247 or e-mail Genetics@michigan.gov

Michigan Resources & Support

Fragile X Association of Michigan
Phone: 313-381-2834
www.fragilex.org/html/michigan.htm

Children's Special Health Care Services
Family Phone Line
Toll-free: 1-800-359-3722
E-mail: ppp@michigan.gov
www.michigan.gov/cshcs

***Early On*® Michigan**
Toll-free: 1-800-EARLY ON
www.1800earlyon.org

Michigan Birth Defects Program
Nurse Follow-up Coordinator
Toll-free: 1-866-852-1247
E-mail: BDRfollowup@michigan.gov

Michigan Genetics Connection
www.migeneticsconnection.org

National Resources & Support

National Fragile X Foundation
Toll-free: 1-800-688-8765
www.fragilex.org/

The Fragile X Research Foundation (FRAXA)
Phone: 978-462-1866
www.fraxa.org/

Family Village
www.familyvillage.wisc

GeneReviews
www.geneclinics.org/profiles/fragilex/

Genetics Home Reference
www.ghr.nlm.nih.gov/condition=fragilexsyndrome

National Institute of Child Health and Human Development
www.nichd.nih.gov/publications/pubs/fragileX/index.htm

Supported in part by project # 6 H91MC00215-04-01 as a Special Project of Regional and National Significance (SPRANS), Title V (as amended), Social Security Act, administered by the Maternal and Child Health Bureau, Health Resources and Services Administration, United States Department of Health and Human Services.
Rev. 4/06